

Importance of clinical examination in the recognition of Herlyn-Werner-Wunderlich Syndrome: case report

A importância do exame clínico no reconhecimento da síndrome de Herlyn-Werner-Wunderlich: relato de caso

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ABSTRACT: *Introduction:* Herlyn-Werner-Wunderlich syndrome (HWWS) is a rare congenital disease of the Mullerian ducts, in which there was a didelphys uterus, hemivagina obstructed by the septum, and ipsilateral renal agenesis. The most common clinical presentation is progressive pelvic pain, dysmenorrhea, and a palpable mass. In some cases, the patient may have normal menstruation due to a hemivagina obstruction, which ends up delaying the diagnosis. *Case Report:* Female patient, 13 years old, admitted with complaints of pelvic pain, intermittent and progressive, a year ago, associated with menstrual irregularity. Performed a radiological propaedeutics and the initial gynecological physical examination was suppressed, the patient is misdiagnosed with ovarian cyst torsion and submitted to exploratory laparotomy. In the postoperative period, after reassessing the clinical history and undergoing a gynecological exam, an anatomical abnormality was observed in the region of the vaginal vestibule, compatible with the vaginal septum and hematocolpo. Colpotomy was performed with drainage of approximately 900 ml of chocolate-like blood and marsupialization of the vaginal septum. *Conclusions:* The importance of clinical examination for early diagnostic completion and to avoid unnecessary surgical procedures is noted, especially in the management of rare and intriguing cases such as HWWS.

Keywords: Menstruation disturbances; Dysmenorrhea; Mullerian ducts; Solitary kidney; Physical examination.

RESUMO: *Introdução:* A síndrome de Herlyn-Werner-Wunderlich (SHWW) é uma doença congênita rara dos ductos müllerianos, em que há útero didelfo, obstrução de hemivagina obstruída por septo e agenesia renal ipsilateral. A apresentação clínica mais comum é dor pélvica progressiva, dismenorreia e massa palpável. Em alguns casos a paciente pode apresentar menstruação normal devido a obstrução de apenas uma hemivagina, fato que resulta em atraso no diagnóstico. *Relato do caso:* Paciente, 13 anos, sexo feminino, com queixa de dor pélvica intermitente e progressiva, há um ano, associada a irregularidade menstrual, é atendida em hospital terciário. Realizada propedêutica radiológica armada e suprimido o exame físico ginecológico inicial, paciente é erroneamente diagnosticada com torção de cisto ovariano e submetida a laparotomia exploradora. Em pós-operatório, após reavaliação da história clínica e realização de exame ginecológico, evidenciou-se uma anormalidade anatômica em região de vestibulo vaginal, compatível com septo vaginal e hematocolpo. Realizada colpotomia com drenagem de aproximadamente 900 ml de sangue de aspecto achocolatado e marsupialização do septo vaginal. *Conclusões:* Denota-se a importância do exame clínico para conclusão diagnóstica precoce e para evitar procedimentos cirúrgicos desnecessários, em especial na conduta de casos raros e intrigantes como a SHWW.

Palavras-chave: Distúrbios menstruais, Dismenorreia, Ductos paramesonéfricos, Rim único, Exame físico.

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INTRODUCTION

Herlyn-Werner-Wunderlich syndrome (HWWS) has a limited understood etiology, with an estimated prevalence of less than 1/1,000,000. It is also known as OHVIRA for its acronyms for obstructed hemivagina and ipsilateral renal agenesis. The most frequent clinical symptoms of the disease are recurrent pelvic pain, severe dysmenorrhea, and a palpable mass, which tends to appear after menacme; rarely, the complaint may be primary infertility¹.

The diagnosis of HWWS requires radiological evidence of uterus didelphys, vaginal septum, and ipsilateral renal agenesis, which is often associated with hematocolpos and hematometra. Surgical treatment must be performed early to relieve symptoms and prevent long-term complications related to retrograde menstrual flow, including piocolpo, endometriosis, and pelvic adhesions².

In most cases, HWWS is not initially recognized, resulting in misdiagnosis and inadequate treatments. Although the diagnosis is sealed with imaging exams, suspicion through a well-performed clinical exam is essential to reduce the time of its identification and institution of an appropriate approach.

This report presents a case of a patient with a syndrome that, although rare, could have been recognized early in referral services, in which the lack of an adequate clinical approach resulted in a late diagnosis and unnecessary surgical procedure. The images presented were authorized by the mother, responsible for the patient, by means of a free and informed consent term, respecting the absolute confidentiality of her identity.

CASE REPORT

Female patient, 13 years old, brown, admitted with complaints of pelvic pain, mainly on the left, intense and progressive, which started after menarche at 12 years old. She denied having sexual intercourse and reported menstrual irregularity. On physical examination, the patient was in regular general condition, eupneic, normotensive. with severe abdominal pain on deep

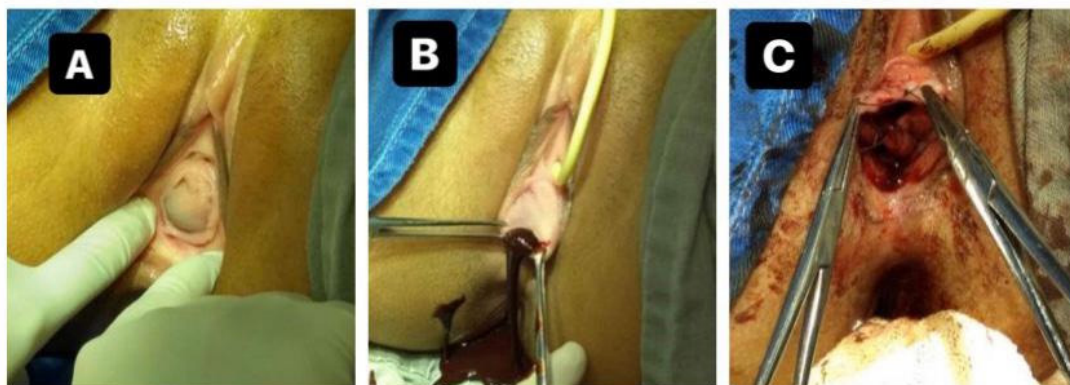
hypogastric palpation and the presence of a palpable mass in the left iliac fossa.

In the clinical history, a patient-reported previous hospitalizations in other services due to the same pain and presented a contrasted tomography of the entire abdomen (performed 42 days before admission). She also had an ultrasound of the lower abdomen (28 days before the date). Both reports demonstrated an expansive pelvic formation in the left adnexal topography, hypoechogenic/hypodense with precise limits of 7x7.4x7.4cm in their largest diameters. In the report of the total abdominal tomography, there was also a description of left renal agenesis. The other structures evaluated did not present pathological changes.

After diagnosing an ovarian cyst torsion hypothesis, the patient was immediately referred to the operating room for an exploratory laparotomy, in which only bladder distention was noted in the cavity inventory. After the procedure, the patient went to the infirmary for surgical recovery and further diagnostic investigation, due to the negative finding in the intraoperative period in view of the suggested initial hypothesis.

On the first postoperative day, after reassessment of the clinical history, the patient's gynecological exam was performed for the first time, in which an anatomical abnormality was found in the region of the vaginal vestibule, compatible with the vaginal septum associated with hematocolpo. On the next day (2 days post-surgery), the patient underwent a new intervention, in which a colpotomy was performed with drainage of approximately 900 ml of chocolate-like fluid and marsupialization of the vaginal septum (Figure 1).

In a brief review of the exams, renal agenesis on the left corroborated the hypothesis of HWWS as the origin of the hematocolpo. However, only a magnetic resonance imaging of the pelvis did evidence the presence of a didelphys uterus (Figure 2). With the results, the patient closed the clinical and radiological criteria for HWWS and, due to the good clinical evolution, she was discharged two days after the surgical correction in good general condition, with due orientation on follow-up, return, and doubts about this rare syndrome.



Source: the authors.

Figure 1: A) Aspect of the vulvar vestibule before surgical correction; B) Colpotomy for drainage of hematocolpos; C) Result of marsupialization of the septum in the left vagina.



Source: the authors.

Figure 2: Magnetic resonance imaging showing a didelphys uterus (arrow) in axial contrast-enhanced.

DISCUSSION

The syndrome's alterations were initially reported by the English surgeon EC Purslow in 1922, but its complete description was only carried out in 1976 by surgeons Herlyn, Werner and Wunderlich who described it as a congenital disease of the Mullerian ducts, in which there was a didelphys uterus, obstructed hemivagina and unilateral renal agenesis^{3,4}. It is estimated that it corresponds to 3.8% of congenital genital malformations⁵.

HWWS occurs from a malformation involving the urogenital system during embryogenesis. The mesonephric ducts (also known as Wolff's ducts) originate the kidney and ureter in the female sex, and has a common mesodermal origin with the paramesonephric ducts (or Muller's ducts), explaining the association often found between genital and urinary tract abnormalities^{6,7}. The defect at the moment of the merger of the Mullerian ducts occurs around the 8th week of embryogenesis and leads to uterine changes and vaginal septa^{8,9}.

In 72.4% of cases, the classic form is presented - uterus didelphys, hemivagina obstructed by the septum and ipsilateral renal agenesis -, but many variations in the anatomical structures involved are recognized⁸. Classification for the HWWS suggests division according to the shape of the vaginal septa in type I: unperforated diagonal septum, type II: perforated oblique septum, and type III: unperforated diagonal septum with cervical

fistula^{2,5,9}. Type I is the most frequent and also observed in the case presented in the current report.

Clinical manifestations appear only after menarche and coincide with the menstrual cycle, which can be identified by progressive dysmenorrhea, painful suprapubic mass secondary to hematocolpos, hematometra, and hematosalpinx^{6,9}. In rare cases, the impounded blood can be infected, generating the condition of piocolpo, pelvic inflammatory disease, and tube-ovarian abscess⁹. In these cases, fever, chills, nausea and vomiting may be present and even progress to septic shock. Other possible complications are endometriosis, infertility and adhesion formation^{1,6,10}.

The diagnostic approach is initiated by anamnesis, and physical examination performed adequately. The hemivaginal septum can be seen through bimanual touch or specular examination. In patients without sexual activity, the vaginal examination can become a difficulty, and digital rectal examination can be used to observe vaginal bulging^{1,5,6,9,10}. However, in symptomatic patients, vaginal inspection is often sufficient to lead to diagnostic suspicion, as it may reveal a vaginal bulge next to a healthy vagina due to hematocolpos¹, as shown in Figure 1.A of the present report.

The investigation proceeds with a pelvic ultrasound, an excellent test for being low-cost, fast, non-invasive, and able to assess the genital system, being, therefore, the most used test initially. However, the examination field is small, the resolution of soft tissues is low and the diagnostic accuracy is examiner dependent⁹. The examination may indicate didelphis or bicornuate uterus, hematocolpos, and renal agenesis⁵. In the case presented, the ultrasound showed only the left adnexal mass, not distinguishing the hematocolpos or the didelphys uterus, suggesting endometrioma or mucinous cystadenoma. This fact contributed to the formulation of the mistaken hypothesis of ovarian torsion.

Computed tomography can also diagnose the disease, but the evaluation of the uterine structure is limited, in addition to being a source of irradiation⁹. Magnetic resonance imaging is the gold standard exam, as it presents greater sensitivity in verifying the vaginal septum, which is essential for surgical planning^{6,9}. The primary differential diagnosis is the transverse vaginal septum, with congenital vaginal atresia being another differential diagnosis, all identified and differentiated by magnetic resonance imaging⁹.

In a study with 70 cases of HWWS, the mean age at onset of symptoms was 17 years, while the mean age at diagnosis was 21 years¹⁰. Clinical manifestations begin 12 to 20 months after menarche, but later, presentations may appear in patients with type II HWWS when there are perforations in the vaginal septum^{6,7,10}.

The diagnosis delay stems from the fact that even with an obstructed hemivagina, patients with HWWS can

present a typical menstrual pattern due to the intact vaginal side, and depending on the shape of the vaginal septum. In addition, when it comes to dysmenorrhea, many doctors generally prefer prescriptions based on anti-inflammatory drugs and oral contraceptives that can suppress or inhibit the menstrual cycle and much of the symptoms⁶.

In the case presented, the patient was diagnosed with HWWS at 13 years of age, only after exploratory laparotomy indicated because it is believed that there was torsion of the ovarian cyst. Unfortunately, 92.9% of patients receive incorrect diagnoses of other conditions, including pelvic mass, ovarian cyst, vaginal cyst with or without infection, teratoma, among others⁷. Misdiagnosis, so common in HWWS, may not be due to the lack of resources for complementary exams, but to the absence of adequate clinical examination, with basic anamnesis and physical examination.

The patient presented brought an ultrasound and abdominal tomography to the referral service, and both did not describe the hematocolpos and the didelphys uterus. They defined cystic expansion in left adnexal topography, in addition to left renal agenesis. The absence of an initial physical examination led to an unnecessary surgical approach, causing morbidity and risks to the patient's life. Many cases of HWWS undergo unnecessary surgery before the definitive diagnosis and treatment, reaching 12.5% in study¹.

The clinical examination has always been one of the essential pillars in medical practice aiming at a more accurate diagnosis, however, this exercise has lost its space in the last decades. More and more, the modern doctor surrenders to the advancement of armed propaedeutic, forgetting basic precepts of the clinical examination, a fact that, in the current medical scenario, results in significant problems. Disregarding semiological principles at the expense of complementary exams can lead to misdiagnosis and iatrogenic.

Diagnosis and intervention should be performed

as early as possible to avoid complications such as pelvic inflammatory disease, piocolpo, endometriosis and infertility^{1,2}. The definitive treatment of HWWS is a surgical approach, to perform oblique excision or marsupialization of the hemivagina septum, drain the hematocolpo, and hematometra, leading to rapid relief of symptoms^{1,2,5}. Vaginoplasty is performed with the help of a speculum or vaginal retractors. In virgin patients, the choice is for resection via hysteroscopy, where the integrity of the hymen is maintained^{2,5}.

Research shows a technique known as "No Touch", suitable for teenagers with narrow and underdeveloped vaginas, with an intact hymen. It is a minimally invasive form by vaginoscopy, which reduces the risks of post-surgical complications and, also, pain after surgery². The concern with the maintenance of the hymen is present in some cultures and should be considered when choosing the surgical technique, individualizing each case. In the present report, we opted for a colpotomy with drainage of the hematocolpos and marsupialization of the septum vaginally due to the lack of a video approach.

CONCLUSION

Herlyn-Werner-Wunderlich syndrome is rare but easy to identify with the proper approach and should be suspected in the presence of progressive dysmenorrhea in the first years after menarche, mainly if associated with renal agenesis. The neglect of the clinical examination - anamnesis and physical examination - within the current medical practice results in iatrogenic conduct, even more so concerning HWWS, in which the imaging exams can be a confounding factor, as in the present report. This fact needs more attention from medical professionals, especially gynecologists, for the timely and early recognition of congenital anomalies, in order to avoid complications from inadequate and late diagnoses, as well as unnecessary surgical approaches.

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